ANTE NATAL SCREENING

Birth is a highly technological business these days. From the moment a pregnancy is confirmed, a wide range of tests is available to monitor whether the pregnancy is progressing 'normally'. Some tests are routine, with little or no choice about whether to have them or not. Others are aimed at particular groups of women thought to be 'at risk'. Ante-natal screening raises many questions that should not just be left to the medical profession and the scientific world.

The Politics of Pre-Natal Screening

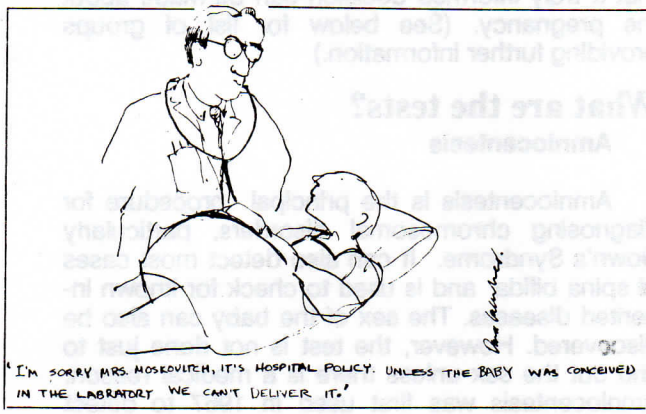
In recent years, the developments in screening techniques have been heralded by scientists and the medical profession as major advances. Doctors and politicians as well as consumer organisations have demanded the right of every woman to have access to pre-natal screening. We need to look at what role women play in making decisions about screening and question the assumptions that determine who gets screened and how. The two major issues here are informed consent and attitudes about disability.

Are we told what we are being screened for? Are we asked whether we want to have the tests done? What choice do we have in the matter? Are we told that we can choose not to have any of them? How are we treated if we refuse to have a test? Are tests offered on condition or on the assumption that we opt for a termination if a disability is discovered? How much are we told about the nature of the disability being screened for? Is disability counselling biased towards abortion? What qualifications do 'experts' have on what each particular disability means in terms of day to day living? Whose value judgments are operating when assessing the 'usefulness' of a potential life?

Very often, it is assumed that women would opt for an abortion if a birth defect is discovered. One attitude to disability in relation to screening is that all congenital (this simply means present at birth, not necessarily genetic) disabiltity will soon be a thing of the past and that this would be a good thing. Women who resist screening may be seen as irresponsible and be made to feel guilty. Doctors often assume that if an abnormality is discovered, women would automatically opt for an abortion. But late abortion isn't necessarily what women would 'choose'. Screening is not seen as an end in itself, ie. as a source of information to prepare a woman for the birth.

A three year research project into women's experiences of pre- natal screening highlighted some of these issues. The project conclusion states "In the context of a society which devalues people with disabilities and holds mothers to be responsible both for the prevention of congenital abnormality and for the care of children who are born with disabilities, women have looked to prenatal screening as a means of allaying fears about foetal abnormality and of gaining some control over the possibility of producing a baby with a severe handicap. For women considered to be at risk, the technical potential of pre-natal screening for enhancing a woman's reproductive choices and for making pregnancy a more carefree experience is a real one. However, prenatal screening has not been developed with the interests of women primarily in mind, it often fails to take account of women's needs and it has also increased the potential for others to control women's reproduction. Also for many women, particularly women not considered at risk, screening has become yet another way in which medical care detracts from the possibility of pregnancy being experienced as a normal and enjoyable event. Furthermore, far from relieving the responsiblity that is placed on mothers, prenatal screening can be seen as feeding into and reinforcing a dominant ideology that poses solutions to disability in terms of medical science and maternal responsiblity, rather than social and political change.

It is in this context that we provide information about the range of tests that are currently available.



Who is 'at risk'?

There are three groups of women who are considered to be 'at risk' of carrying fetuses with genetic disabilities and may want pre-natal diagnosis. The first is women whose family history (or partner's history) includes a genetic disease or birth defect. The second is women and their partners from certain ethnic groups in which specific inherited diseases occur. The third is women aged 35 and over or women with older partners, as the risk of fetal disability increases with age. Age is an important factor with Down's Syndrome. If you are 25, you have a 1 in 1,500 chance of having a Down's baby, at 30 you have a 1 in 800 chance, at 35 you have a 1 in 300 chance, at 38 you have a 1 in 180 chance, at 40 a 1 in 100 chance and at 45 a 1 in 30 chance.

What can be screened for?

It is important to bear in mind that ante-natal screening cannot reveal all possible birth disorders. Noone knows why the majority of problems occur (things like heart defects or cleft lip) and they can't be detected until birth. Moreover, screening cannot indicate the degree of disability. For example, with Downs Syndrome a baby might only be mildly affected in some cases but severely affected with multiple medical problems in others.

(See glossary for definitions of the following)

- parental dominant gene conditions eg. Huntington's chorea

- recessive gene conditions in which parents are carriers but never manifest the problems themselves, such as Tay-Sachs and sickle cell disease.

- neural tube defects eg. anencephaly (where a part or all of the brain is missing) and spina bifida (caused by a break in the neural tube ie. spine)

- chromosomal anomalies eg. Down's Syndrome, Turner's and Klinefelter's syndrome.

- sex linked diseases eg. Duchennes muscular dystrophy and haemophilia.

What must be kept in mind is that all of these conditions affect babies (and consequently the amount of care they will need) in very different ways. When a woman is tested for them, adequate and sympathetic counselling is essential so that a truly informed decision can be made about the pregnancy. (See below for list of groups providing further information.)

What are the tests?

Amniocentesis

Amniocentesis is the principal procedure for diagnosing chromosomal disorders, particularly Down's Syndrome. It can also detect most cases of spina bifida and is used to check for known inherited diseases. The sex of the baby can also be discovered. However, the test is not done just to find out the sex unless there is a medical reason. Amniocentesis was first used in 1967 to detect chromosomal problems and then extended to neural tube defects in 1973 and further developed since then.

Amniocentesis is offered to women considered to be 'at risk' of carrying a fetus with a birth defect, especially to women 35 or over. The lower age limit is decided by each individual health authority or consultant but it ranges from 35 years of age upwards. In more than 95% of tests, even in high risk groups, results show no disorders. Amniocentesis can only screen for certain conditions. A 'clear' test result is not a guarantee of an unaffected fetus. As mentioned above, not all birth disorders can be detected.

What the test involves:

It is done usually at 16 weeks but no earlier, as an outpatient test and can be done in a morning or afternoon appointment. You do not have to be admitted to hospital. First, an ultrasound scan is done to make sure you are at least 16 weeks pregnant (to ensure there is enough amniotic fluid to be drawn off) and to check the position of the baby and the placenta. You will need a full bladder for this scan. Then your skin is cleaned with antiseptic and a small injection may be given to numb the area. A fine needle is then passed through your abdomen and into the womb and a sample (about 4 teaspoonsful) of the fluid that surrounds the fetus is removed with a syringe. An ultrasound scan is used to guide the needle to the right place and to ensure that it does not touch the placenta. (We have heard of hospitals that do not do the procedure using ultrasound scan; women should ensure that their hospital uses a scan in order to minimise risks.) The sample is then sent to the laboratory for testing. Most women say the test is not painful and that the thought of it was much worse than the actual test. Some women say it feels no more painful than any injection or blood test. You will be able to go home afterwards, though it is a good idea to rest for an hour. Some women may get a 'tightening' feeling in the womb afterwards or may feel a little sore the next day. This is not unusual. You should take things easy for a couple of days and avoid any heavy lifting or stenuous exercise.

The results of the chromosome test, which involves 'growing' the cells from the fluid, take about 3-5 weeks. Sometimes the cells do not grow well and a repeat test is organised immediately. The chromosome test is very accurate. Every single one of the 46 chromosomes that make up a human cell has to be inspected. This can take a long time, as much as half a day. The possibility of a false positive is very low indeed, as low as 1 in every 200,000 tests.

The results for the spina bifida test (done on the same sample of fluid) takes about a week to come through. A raised level of alpha-feto-protein (AFP) in the fluid means an over 99% certainty that an open neural tube defect or other abnormality is present. This test is not as sensitive as the chromosome test, but does pick up about 80 - 90% of affected pregnancies. Some others would be seen on the ultrasound scan.

What are the risks?

The miscarriage rate is currently 1 in 150/200 pregnancies. It is not understood why these miscarriages happen and there is no way of predicting which women are likely to miscarry because of the test. However, a higher AFP level may cause miscarriage. Other complication that has occurred in a further 1% of pregnancies is unexplained breathing difficulties. Every additional insertion carries an additional risk. Very occasionally the amniotic fluid sample is contaminated with blood from the mother and in very rare cases,the mother's cells are analysed, giving a false negative result.

Alpha-Feto-Protein Test

The Alpha-Feto-Protein (AFP) test is carried out at 16 weeks. It is a blood test to discover

how much AFP (protein produced by the by the fetus) is present in your bloodstream. As your pregnancy progresses, the level of AFP in your blood increases. At 16 weeks it is possible to see whether the AFP level is higher than it should be. A raised AFP level could mean that your pregnancy is further on than you realised or that you are carrying more than one baby (an ultrasound scan will check that). However, in some circumstances, it may indicate the presence of a fetus with a neural tube defect (NTD). If the AFP level is raised, you should be given a second test. And if raised levels are still indicated, other tests will be performed to see if the fetus is affected. A 'real time' scanner (giving a continuous picture, rather than a single shot) will often detect a fetal NTD. Older equipment can only tell you whether or not you are having twins and double check your dates. If it is still not clear, amniocentesis is offered and a diagnostic test for the enzyme acetylcholinsterase is done to confirm the NTD.

About 50 out of 1,000 women will have positive AFP tests. The procedure varies according to local policy but usually these women are tested again immediately. After the second test, about 30 will still have excessive amounts of AFP. A subsequent ultrasound will show that about half of these women have more advanced pregnancies, are carrying more than one baby or their babies have died. Of the remaining fifteen who have amniocentesis to check the AFP concentrations in amniotic fluid, one of these women will have an NTD confirmed. (Another study indicated a 1 in 10 chance of NTD confirmation).

In some areas AFP blood tests are given routinely to everyone. Other health authorities use ultrasound more often as this technique is getting more and more sophisticated. In others, AFP tests are only offered if you have a family history of NTD.

Recent research work at St Bartholomew's Hospital in London is highlighting a new use for the AFP test. Whilst a high level of AFPmay indicate an NTD, a low level may indicate a higher risk of Down's Syndrome. Researchers have analysed world data to produce an individual ratings system for each woman, depending on her age and her AFP levels. By looking at the rating, a younger women could be offered amniocentesis whereas an older woman may be seen not to need it. Because of the age restriction, many young women are not screened and women under 35 bear about 80% of babies born with Down's Syndrome (this is because there are far more pregnant women under than over 35.)

Chorionic Villus Sampling (CVS)

CVS is a relatively new technique that is still in research stages. It was pioneered in the Soviet Union, China and Finland. The test can pick up the same conditions as the amniocentesis test, except that it does not detect spina bifida and a small number of rare conditions not routinely screened for (usually known hereditary ones).

The Medical Research Council (MRC)is organising a randomised control trial in 23 centres in Britain and 5 in Europe (Finland, Italy, Switzerland, Holland and Hungary). Canada and Denmark are conducting separate trials. Between 2,000 and 4,000 women will be involved in the MRC trials lasting until about the end of 1988. About 750 women have participated in the tests so far in Britain.* Results from the trial will take about a year to analyse.

In the trial, 50% of women are randomly assigned to have CVS and 50% to have amniocentesis, without being able to choose which test to have. If you are over 35 or run a special risk of genetic or chromosomal problems and are less than 11 weeks pregnant, you can join the study. It is possible to choose CVS if you are a woman considered of proven risk (for example, if you and your partner are sickle cell carriers or if you have already had a baby with a specific problem.) CVS can be done at about 8 - 10 weeks and the results are usually available in 1 - 2 weeks. Early in pregnancy (at 6 weeks) the embryo is encased in a membrane (the chorion) from which hairlike wisps of tissue protrude (villi). By the end of the first trimester (ie. 14 weeks), part of the chorion thickens to become the placenta, the rest becomes a thin membrane and the villi disappear. But between 8 and 12 weeks of pregnancy, doctors can remove tiny plugs of tissue from the villi without, apparently, disturbing the pregnancy. This tissue can then be analysed to check for certain birth defects (in the same way as amniotic fluid is analysed in amniocentesis). Since CVS was first developed, more and more disabilities are being screened for eg. cystic fibrosis can now be detected.

What the test involves:

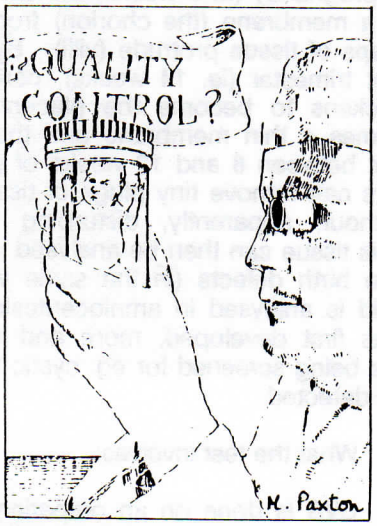
CVS is done on an outpatient basis. You do not have to be admitted to hospital or have an anaesthetic. An ultrasound scan is done first (you need to have a full bladder for this). If it is done through the cervix, your vagina is cleaned with some antiseptic solution and a thin tube is passed through the cervix with the ultrasound scan guiding the tube to the right place. A tiny fragment (about the size of a few grains of rice) from the edge of the chorionic tissue is gently sucked into the tube and removed and sent to the laboratory for tests. Most women say that the test was uncomfortable rather than painful, a bit like having a smear done. If it is done through the abdomen, the same procedure as with amniocentesis is followed. Very occasionally, the cells may not grow well in the laboratory and the test then has to be repeated as soon as possible. It normally takes 10 - 20 minutes to do and you will be able to go home afterwards, preferably after resting for about

an hour. It's a good idea to take it easy for a few days and avoid any heavy lifting or strenuous exercise.

CVS was originally done through the cervix (neck of the womb), but now some hospitals prefer doing it through the abdomen. King's College Hospital maintains that the miscarriage rate is reduced using the transabdominal route, with a higher success rate in obtaining tissue at the first attempt. They point out that it is easier to train staff who are already used to doing amniocentesis.

What are the risks?

The risks of the actual technique are as yet unknown. The more skilled practitioners get, the lower the risk. The miscarriage rate has not been established, but it is estimated to be higher than amniocentesis, possibly 1 in 50. There is a possibility that CVS done transabdominally is safer than when done transcervically. Other areas of concern are: false positive diagnoses, introduction of infection into the uterus (particularly when done transcervically), fetal growth retardation and the risk of exposing the fetus to an unusually large amount of ultrasound early in pregnancy. The current trial should provide information on the real risks involved in this technique. The potential use of CVS as a screening tool is being questioned because many of the fetuses with chromosomal anomalies detected by the tenth week will spontaneously abort.



Ultrasound

Ultrasound is the most common prenatal test. It is used routinely from 12 weeks in a very large number of pregnancies whereas in some areas it is not available. Ultrasound uses high frequency sound waves to form a picture of the fetus on a TV screen.You lie on your back, and a small instrument called a 'transducer' is passed over your belly. Some new machines have a transducer that is inserted into your vagina. Ask the doctor or technician to see the image on the screen and point out which bit is which.

Ultrasound is used to determine gestational age and to indentify multiple pregnancies. It establishes the position of the placenta and any growths or abnormalities in the shape of your womb which may obstruct delivery. It can sometimes determine the sex of the baby. It can also pick up physical abnormalities in a fetus of 16 weeks and fetal death. It can sometimes detect closed NTDs and one or two conditions that severely damage the brain or kidney of a fetus. In later pregnancy, the scan can spot potential problems such as placenta praevia (when the placenta covers the neck of the womb).

In some hospitals, 'real time' scanning is used ie. a continuous picture rather than a single shot. This can be so accurate that it may be preferred to amniocentesis when neural tube defects are suspected. It has the advantage of instant results, no risk of miscarriage and more detailed information. However, such accurate technology is not widely available and requires highly trained staff. The possible risk of prolonged exposure to ultrasound should not be overlooked.

What are the risks?

There is no evidence as yet that the procedure is harmful at this stage of a pregnancy (ie. 12 weeks and on) However, there have been suggestions that ultrasound should not be used routinely during pregnancy unless a woman is considered to be 'at risk'. It has further been suggested that ultrasound could be harmful during the early stages of pregnancy, ie. in the initial few weeks. It is possible that some future unforeseen problems may emerge. However, there are no plans to research the long term effects of ultrasound. Studies in the United States have shown that ultrasound can damage the chromosomes in animals and can alter the way cells move when they are grown outside the body in tissue culture. The World Health Organisation, the Dainish Health Ministry and the American Food and Drug Administration all advise that it should only be used when there is a medical reason to suspect that something might be wrong with the pregnancy.

Fetoscopy

Fetoscopy is a general term covering a number of procedures which involve direct examination of the fetus or fetal blood or tissue. It is a new area of screening. A fetoscope, a fine fibre-optic telescope, is passed through the abdomen into the womb so that the fetus can be seen. It is performed in hospital under local anaesthetic between 16 and 22 weeks. It is used to detect Sickle Cell (although CVS will probably be used more and more for this) and haemophilia. There are risks attached: miscarriage, uterine bleeding, infection, puncture of other organs, rupture of fetal blood vessels, damage to the eyes (due to the strong light used), leakage of fetal blood into the mother's circulation and psychological stress and anxiety for the woman. It is not widely available as yet, but fetoscopy may be used to perform surgical operations on a fetus while it is still in the womb.

*These are some of the hospitals are taking part in CVS trials: Birmingham Maternity Hospital; Bolton General Hospital; University Hospital of Wales, Cardiff; Western General Hospital, Edinburgh; Duncan Gurhrie Institute of Medical Genetics, Glasgow; Perivale Maternity Hospital, Greenford; Guy's Hospital, London; University College Hosptial, London; Luton and Dunstable Hospital, Luton; Princess Mary Maternity Hospital, Newcastle upon Tyne; John Radcliffe Hospital, Oxford; Solihull Maternity Hospital.

Groups to contact for more information:

Association for Spina Bifida and Hydrocephalus 22 Upper Woburn Place, London WC1H 0EP 01-388-1382

CompassionateFriends(international bereaved parents org.)5 Lower Clifton HillClifton, Bristol BF8 1BT 0272-292-778

Contact a Family with a Handicapped Child (mutual support) 16 Strutton Ground, London SW1P 2HP 01-222-3969

Down's Children Association 3rd Flr. Entrance Through Hornes, 4 Oxford Street, London W1N 9FL 01-580-0511/2

Royal Society for Mentally Handicapped Children and Adults (MENCAP) 123 Golden Lane, London EC1Y 0RT 01-253-9433

Support After Termination for Abnormality (SAFTA) National Office: 22 Upper Woburn Place, London WC1H 0EP 01-267-9067

References

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Infection and CVS, Brambati and Varotto, Lancet 14.9.87

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GLOSSARY

Chromosomal Disorders

Down's Syndrome - caused by an error in chromosome formation, causing varying levels of disability; it is associated with congenital heart disease and decreased life expectancy.

Klinefelter Syndrome - due to an extra X chromosome (the sex chromosome); occurs 1 in 700 live births, causing female characteristics in males. May cause infertility and mild learning disability.

Turner's Syndrome - another sex chromosome related disorder occurring in 1 in 2500 female babies; causes underdeveloped female characteristics eg. ovaries don't develop and menstruation does not occur.

Sex Linked Disorders

Duchenne Muscular Dystrophy - occurs 1 in 3000 boys, causing progressive muscular weakness and early death in 1/3 of cases.

Haemophilia - occurs only in boys and causes impaired blood clotting and a tendancy to bleed easily.

Metabolic Disorders

Cystic Fibrosis - occurs in 1 in 2000 births, causing the mucus glands to secrete abnormally thick mucus and the sweat glands very salty sweat. Can cause chronic lung, bowel and liver disease.

Huntington's Chorea - a degenerative disease of the central nervous system which develops some time in mid-life; about 60 new cases per year in England and Wales.

Tay Sachs Disease - usually occurs in those of Ashkenazi Jewish descent, caused by a deficient enzyme; usually develops within the first 6 months with progressive neurological degeneration.

Genetic Blood Disorders

Sickle Cell Disease - a group of blood conditions which included sickle cell anaemia, haemoglobin SC disease and sickle beta- thalassaemia. The most common is sickle cell anaemia which is due to a problem with the haemoglobin structure, causing red blood cells to clump under certain circumstances. It mainly affects people of Afro-caribbean origin but occurs in others as well.

Multifactorial Disorders

Neural Tube Defects (NTD) - occurs in 1-8 per 1000 live births. It can be divided into two types:

I. Anecephaly - is due to absence of the upper brain; it usually results in still birth but a small number of live births have survived for a few days.

2. Spina bifida - a defect of the spinal tube causing the nerves of the spinal column to be exposed in 85% of cases; the defect can also be closed and can cause varying degrees of paralysis and hydrocephaly (fluid on the brain).

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